



Fig. 1

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Table 1

Number and Frequency (%) of ACTN3 Genotypes and Frequency (%) of ACTN3 Alleles in Controls and Elite Sprint/Power and Endurance Athletes

GROUP (n)	No. (%) WITH GENOTYPE			ALLELE FREQUENCY (%)	
	RR	RX	XX	R	X
Male:					
Controls (134)	40 (30)	73 (54)	21 (16)	57	43
Sprint (72)	38 (53)	28 (39)	6 (8)	72	28
Endurance (122)	34 (28)	63 (52)	25 (20)	54	46
Female:					
Controls (292)	88 (30)	147 (50)	57 (20)	55	45
Sprint (35)	15 (43)	20 (57)	0 (0)	71	29
Endurance (72)	26 (36)	25 (35)	21 (29)	53	47
Total:					
Controls (436)	130 (30)	226 (52)	80 (18)	56	44
Sprint (107)	53 (50)	48 (45)	6 (6)	72	28
Endurance (194)	60 (31)	88 (45)	46 (24)	54	46

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TABLE 2: Genotyping of R577X in ACTN3 in Caucasians Elite Athletes.

Strength	Sport	ID	Sport Institute	Total Number	577RR (%)	577RX (%)	577XX (%)
Endurance	Rower	RT492	AIS	64	22	28	14
		- RT556			(34.4%)	(43.8%)	(21.8%)
Endurance	Triathlete	RT977	AIS	13	3	8	2
		- RT989			(23.1%)	(61.5%)	(15.4%)
Endurance	Cyclist	RT990	AIS	9	4	2	3
		- RT998			(44.4%)	(22.2%)	(33.3%)
Endurance	Track Cyclist	KN246	AIS	22	7	7	8
		- KN275			(31.8%)	(31.8%)	(36.4%)
Endurance	Marathon	KN310	AIS	1	0	0	1
Endurance	All above		AIS	108	36 (33.3%)	45 (41.7%)	27 (25.0%)
Sprint	Swimmer	RT901	AIS	45	17	25	3
		- RT1018			(37.8%)	(55.6%)	(6.6%)
Sprint	Track Cyclist	KN246	AIS	8	4	3	1
		- KN275			(50.0%)	(37.5%)	(12.5%)
Sprint	Athletics	KN276	AIS	30	16	13	1
		- KN309			(53.3%)	(43.3%)	(3.3%)
Sprint	All above		AIS	83	37 (44.6%)	41 (49.4%)	5 (6.0%)
Africa Zulu				88	69 (78.4%)	18 (20.5%)	1 (1.1%)
Australian Caucasian Control				152	46 (30.0%)	78 (52.0%)	28 (18%)

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Table 3 SNPs identified in the ACTN3 gene to date
NCBI SNP CLUSTER ID

rs2229456
rs2229455
rs2229454
rs2228325
rs1126675
rs7949754
rs7924602
rs5792393
rs4990284
rs4990283
rs4013815
rs3937320
rs3837428
rs3814736
rs3814735
rs3782080
rs2511217
rs2511216
rs2509559
rs2509558
rs2305537
rs2305534
rs2290463
rs2275998
rs2096583
rs2000939
rs1815739
rs1791690
rs1671064

10/527 831

rs1188610
rs679228
rs678397
rs677488
rs647476
rs647029
rs618838
rs607736
rs597626
rs544021
rs540874
rs538330
rs531490
rs509556
rs490998
rs13897
rs4576
rs1189338
rs1201433
rs640213
rs3737525
rs3178740
rs3180065
rs3180064
rs3180063
rs3867132
rs608504
rs610293
rs3825065

www.ncbi.nlm.nih.gov/SNP/snp_ref

TSC: The SNP Consortium website

TABLE 4. Symbols, full names, and cytogenic location of nuclear and mitochondrial genes of the 2002 Human Gene Map for Performance and Health-Related Fitness Phenotypes.

Gene or Locus Name Location

A B

ACADVL Acyl coenzyme A dehydrogenase, very long chain 17p13-p11
ACE Angiotensin I converting enzyme 17q23
ADRA2A Alpha-2A-adrenergic receptor 10q24-q26
ADRB1 Adrenergic, beta-1-, receptor 10q24-q26
ADRB2 Beta-2-adrenergic receptor 5q31-q32
ADRB3 Beta-3-adrenergic receptor 8p12-p11.2
AGT Angiotensinogen 1q42-q43
ANG Angiogenin, ribonuclease, RNase A family, 5 14q11.1-q11.2
APOE Apolipoprotein E 19q13.2
ATP1A2 ATPase, Na₂/K₂ transporting, alpha-2 polypeptide 1q21-q23
ATP1B1 ATPase, Na₂/K₂ transporting, beta 1 polypeptide 1q22-q25
BDKRB2 Bradykinin receptor B2 14q32.1-q32.2

C D E F G

CASO2 Calsequestrin 2 (cardiac muscle) 1p13.3-p11
CFTR Cystic fibrosis transmembrane conductance regulator, ATP-binding cassette (sub-family C, member 7) 7q31.2
CKM Creatine kinase, muscle 19q13.2-q13.3
CNTF Ciliary neurotrophic factor 11q12.2
CPT2 Carnitine palmitoyltransferase 2 1p32
COL1A1 Collagen, type I, alpha 1 17q21.3-q22.1
EDN1 Endothelin 1 6p24.1
ENO3 Enolase 3, (beta, muscle) 17pter-p11
FABP2 Fatty acid binding protein 2 4q28-q31
FGA Fibrinogen, A alpha polypeptide 4q28
FGB Fibrinogen, B beta polypeptide 4q28
GDF8 (MSTN) Growth differentiation factor 8 (myostatin) 2q32.2
GNB3 Guanine nucleotide binding protein (G protein), beta polypeptide 3 12p13

H I K L M

HLA-A Major histocompatibility complex, class I, A 6p21.3
HP Haptoglobin 16q22.1
IGF1 Insulin-like growth factor I 12q22-q23
IGF2 Insulin-like growth factor 2 11p15.5
IL-6 Interleukin-6
KCNQ1 K₂ voltage-gated channel, KQT-like subfamily, member 1 11p15.5
LDHA Lactate dehydrogenase A 11p15.4
LPL Lipoprotein lipase 8p22
MTCO1 Cytochrome c oxidase I mtDNA 5904-7445
MTCO3 Cytochrome c oxidase III mtDNA 9207-9990
MTCYB Cytochrome b mtDNA 14747-15887
MTND1 NADH dehydrogenase 1 mtDNA 3307-4262
MTND4 NADH dehydrogenase 4 mtDNA 10760-12137

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MTND5 NADH dehydrogenase 5 mtDNA 12337-14148
MTTE Transfer RNA, mitochondrial, glutamic acid mtDNA 14674-14742
MTTI Transfer RNA, mitochondrial, isoleucine mtDNA 4263-4331
MTTK Transfer RNA, mitochondrial, lysine mtDNA 8295-8364
MTTL1 Transfer RNA, mitochondrial, leucine 1 (UUR) mtDNA 3230-3304
MTTL2 Transfer RNA, mitochondrial, leucine 2 (CUN) mtDNA 12266-12336
MTTM Transfer RNA, mitochondrial, methionine mtDNA 4402-4469
MTTT Transfer RNA, mitochondrial, threonine mtDNA 15888-15953
MTTY Transfer RNA, mitochondrial, tyrosine mtDNA 5826-5891
MyHC myosin Heavy-chain
N O P Q R S T U V
NOS3 Nitric oxide synthase 3 (endothelial cell) 7q36
NPY Neuropeptide Y 7p15.1
PAI1 Plasminogen activator inhibitor 1 7q21.3-q22
PFKM Phosphofructokinase, muscle 12q13.3
PGAM2 Phosphoglycerate mutase 2 (muscle) 7p13-p12
PGK1 Phosphoglycerate kinase 1 Xq13
PHKA1 Phosphorylase kinase, alpha 1 (muscle) Xq12-q13
PON1 Paraoxonase 1 7q21.3
PPARA Peroxisome proliferative activated receptor, alpha 22q13.31
PPARG Peroxisome proliferative activated receptor, gamma 3p25
PYGM Phosphorylase, glycogen, muscle 11q12-q13.2
RYR2 Ryanodine receptor 2 (cardiac) 1q42.1-q43
SGCA Sarcoglycan, alpha (50kDa dystrophin-associated glycoprotein) 17q21
S100A1 S100 calcium binding protein A1 1q21
SUR Sulfonylurea receptor 11p15.1
TGFB1 Transforming growth factor beta 1 19q13.2
UCP2 Uncoupling protein 2 11q13
UCP3 Uncoupling protein 3 11q13
VDR Vitamin D (1,25-dihydroxyvitamin D3) receptor 12q12-q14

The gene symbols, names and cytogenetic locations are from the Locus Link web site (<http://www.ncbi.nlm.nih.gov/LocusLink>) available from the National Center for Biotechnology

Information (NCBI). For mitochondrial DNA, locations are from the human mitochondrial genome data base (<http://www.mitomap.org>).

TABLE 5. Endurance phenotypes and case-control studies (DNA polymorphisms).

Gene	Location	Athletes			N	Controls	
		N	Sports	Freq.		Freq.	P
ADRA2A	10q24-q26	140	Endurance	6.7/6.7: 0.77	141	6.7/6.7: 0.62	0.037
				6.7/6.3: 0.21		6.7/6.3: 0.34	
				6.3/6.3: 0.02		6.3/6.3: 0.04	
				6.7: 0.88		6.7: 0.8	
				6.3: 0.12		6.3: 0.2	
ACE	17q23	64	Endurance	II: 0.30	118	II: 0.18	0.03
				ID: 0.55		ID: 0.51	
				DD: 0.16		DD: 0.32	
				I: 0.57	Ref.	I: 0.43	0.02
				D: 0.43		D: 0.57	
		79	Running	I: 0.57		I: 0.49	0.039
				D: 0.43	Pop.	D: 0.51	
		25	Mountain-eering	NA	Ref.	NA	
					Pop.		0.003
		60	Elite athelets (cycling, running, handball)	II: 0.25	Ref.	II: 0.16	0.0009
				ID: 0.58	Pop.	ID: 0.45	0.004
				DD: 0.17		DD: 0.39	
				I: 0.54		I: 0.38	
				D: 0.46	1248	D: 0.62	
		56	Elite swimmers (subsample of 103 swimmers)	II: 0.15		II: 0.24	
				ID: 0.39		ID: 0.49	
				DD: 0.46		DD: 0.27	
				I: 0.34		I: 0.48	
				D: 0.66		D: 0.52	

Reference: Perusse et al. 2003 "The human gene map for performance and health-related fitness phenotypes: the 2002 update" Med. Sci. Sports Exerc. 35: 1248-1264.

TABLE 6. Genotype and allelic frequencies of *ACTN3* 577R/X alleles in human populations.

Ethnic group	No. of chromosomes	No. of genotypes		Relative allele frequency of 577X
		RX	XX	
Asian	56	14	7	0.5 \pm 0.07
Javanese	96	28	12	0.54 \pm 0.05
Native American	14	2	2	0.43 \pm 0.14
Asia/Americas	166	44	21	0.52 \pm 0.04
Hispanic	64	16	5	0.41 \pm 0.06
White	214	47	21	0.42 \pm 0.03
Europe	278	63	26	0.41 \pm 0.03
Aboriginal	174	33	9	0.29 \pm 0.03
Australian				
PNG Highlander	78	16	6	0.36 \pm 0.05
Australasia	252	49	15	0.31 \pm 0.03
African American	90	12	6	0.27 \pm 0.05
African Bantu	156	14	1	0.10 \pm 0.05
Africa	246	56	7	0.16 \pm 0.05
Unknown	152	50	11	0.47
Total	1094	232	80	